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REVERSAL OF HEART FAILURE AFTER USE OF SIROLIMUS IN A FETAL RHABDOMYOMA: CASE REPORT

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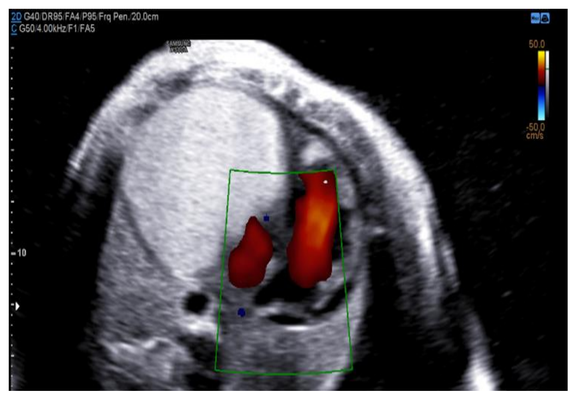
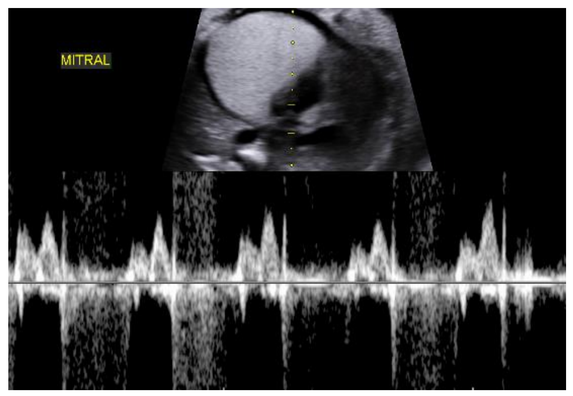
OBJECTIVE

To report a successful case of a large rhabdomyoma treated with sirolimus that was diagnosed in the third trimester of pregnancy during a routine ultrasound and review the literature on the use of sirolimus in fetuses with symptomatic rhabdomyomas.

METHODS

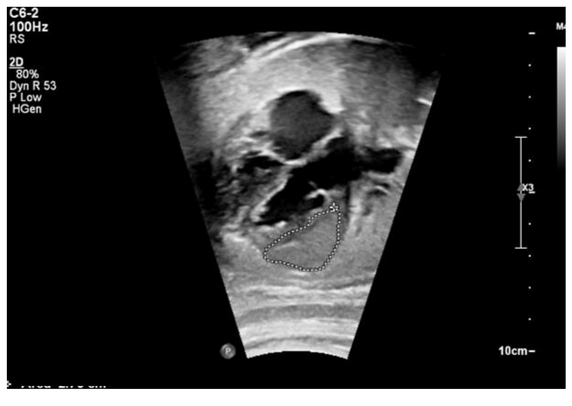
A 37-years old primigravida was referred to our unit of fetal medicine at 29 weeks and 4 days due to the diagnosis of fetal cardiac tumor associated with heart dysfunction and polyhydramnios. The cardiac morphology was anatomically normal except for a **homogeneous echogenic nodule, with no Doppler velocimetry flow, adhered externally to the left ventricle**, measuring 39,2 x 31,7 mm (area of 12.6cm^2), **corresponding to 40% of the thoracic area**. Neurossonography and fetal magnetic resonance were performed, and after that sirolimus therapy and serial fetal evaluation were started. The patient was hospitalized during treatment and oral sirolimus was initiated at a dose of 4 mg/day and was progressive increased up to 8 mg/day. The goal was to achieve the serum concentration between 5 and 10 ng/ml. During the treatment, we analyzed serial blood to control the levels of sirolimus, blood cell count, liver enzymes, cholesterol and triglycerides and we performed serial ultrasound scans and cardiovascular profile score to assess the risk of progression to hydrops.

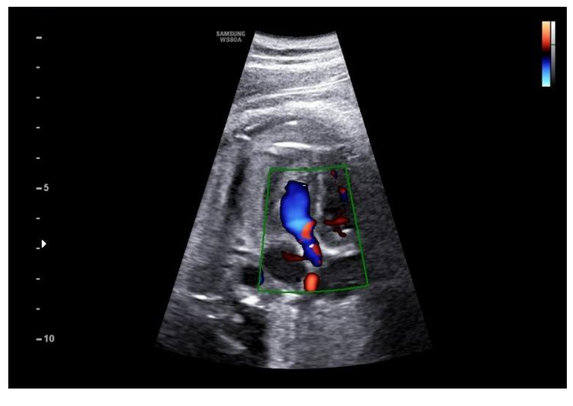




RESULTS

Fetal morphological evaluation demonstrated hyperechogenic subependymal node under the anterior horn of the left lateral ventricle and at least two others small hyperechogenic images in the brain parenchyma. Those findings were also confirmed by fetal magnetic resonance imaging and the patient was advised about the diagnosis of tuberous sclerosis. After sirolimus therapy, we observed progressive tumor reduction and resolution of fetal cardiac dysfunction**. The echocardiography performed at 37 weeks and 3 days demonstrated approximately 50% reduction in the tumor area compared to the first evaluation** (32,0 x 22, 0 mm - area 6,3cm^2). **In addition, the left cardiac chambers had normal dimensions and there was no cardiac dysfunction.** Regarding the side effects of sirolimus, the patient presented hypertriglyceridemia above 500mg/dL from day 28 of treatment. On day 48, the triglycerides reached levels of severe hypertriglyceridemia (above 880mg/dL) and the dose of administration was reduced. In order to minimize the perioperative immunosuppression, the drug was discontinued one week prior to the delivery. The patient delivered a male infant at 38 weeks and 2 days of pregnancy by cesarean section. Postnatal echocardiography demonstrated one hyperechogenic nodule in the apical region of both ventricles, measuring 30,0 x 15,0 mm, with no obstruction of flow. There were normal right chambers and slightly enlarged left chambers. Both ventricles had normal function. The electroencephalography did not show convulsion episodes and the newborn was discharged 5 days after birth. The echocardiography with 3 months old indicates the maintenance of the cardiac tumor on the left ventricle, measuring 34,0 x 104,0 mm, without heart dysfunction. Café au lait spots appeared on the skin, and the infant maintains follow up with pediatric cardiology and neurology and has a good neuropsychomotor development until now.





CONCLUSION

This case adds up to the few cases on international literature and suggesting that oral sirolimus is a good therapeutic option to treat symptomatic fetal rhabdomyomas. However, it is mandatory that multicenter studies try to gather data for better describe prognosis of rhabdomyomas with cardiac disfunction and its treatment, and to determine the efficacy and safety of treatment during pregnancy. It's also important, when treatment is indicated, to close follow the mother concerning triglycerides levels.